



TBC1D24 gene

TBC1 domain family member 24

Normal Function

The *TBC1D24* gene provides instructions for making a protein whose specific function in the cell is unclear. Studies suggest the protein may have several roles in cells. The TBC1D24 protein belongs to a group of proteins that are involved in the movement (transport) of vesicles, which are small sac-like structures that transport proteins and other materials within cells. Research suggests that the TBC1D24 protein may also help cells respond to oxidative stress. Oxidative stress occurs when unstable molecules called free radicals accumulate to levels that can damage or kill cells. Studies indicate that the TBC1D24 protein is active in a variety of organs and tissues; it is particularly active in the brain and likely plays an important role in normal brain development. The TBC1D24 protein is also active in specialized structures called stereocilia. In the inner ear, stereocilia project from certain cells called hair cells. The stereocilia bend in response to sound waves, which is critical for converting sound waves to nerve impulses.

Health Conditions Related to Genetic Changes

DOORS syndrome

At least 10 mutations in the *TBC1D24* gene have been identified in people with DOORS syndrome, a disorder involving multiple abnormalities that are present from birth (congenital). "DOORS" is an abbreviation for the major features of the disorder including deafness; short or absent nails (onychodystrophy); short fingers and toes (osteodystrophy); developmental delay and intellectual disability (previously called mental retardation); and seizures. Some people with DOORS syndrome do not have all of these features.

Most of the *TBC1D24* gene mutations that cause DOORS syndrome change single protein building blocks (amino acids) in the TBC1D24 protein sequence. These mutations are thought to reduce or eliminate the function of the TBC1D24 protein, but the specific mechanism by which loss of TBC1D24 function leads to the signs and symptoms of DOORS syndrome is not well understood.

malignant migrating partial seizures of infancy

nonsyndromic hearing loss

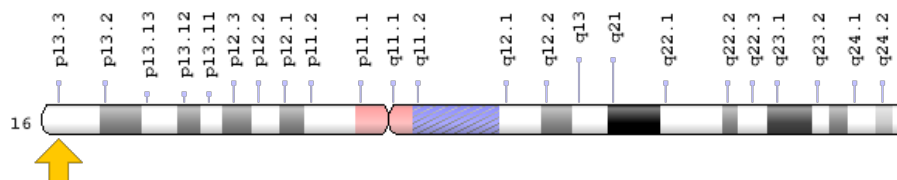
other disorders

TBC1D24 gene mutations have also been identified in people with other seizure disorders, including familial infantile myoclonic epilepsy (FIME), progressive myoclonus epilepsy (PME), and a form of early-infantile epileptic encephalopathy (EIEE16; also called malignant migrating partial seizures of infancy 16). These mutations likely result in impairment of TBC1D24 protein functions related to the development of the brain, but the specific connection between the mutations and these disorders is unclear.

Chromosomal Location

Cytogenetic Location: 16p13.3, which is the short (p) arm of chromosome 16 at position 13.3

Molecular Location: base pairs 2,475,104 to 2,509,669 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DFNA65
- KIAA1171
- skywalker homolog
- TBC/LysM-associated domain containing 6
- TBC1 domain family member 24 isoform 1
- TBC1 domain family member 24 isoform 2
- TLDC6

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Regulation and Coordination of Intracellular Trafficking
<https://www.ncbi.nlm.nih.gov/books/NBK7285/>

GeneReviews

- TBC1D24-Related Disorders
<https://www.ncbi.nlm.nih.gov/books/NBK274566>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TBC1D24%5BTIAB%5D%29+OR+%28TBC1+domain+family,+member+24%5BTIAB%5D%29%29+OR+%28%28DFNA65%5BTIAB%5D%29+OR+%28DFNB86%5BTIAB%5D%29+OR+%28DOORS%5BTIAB%5D%29+OR+%28EIEE16%5BTIAB%5D%29+OR+%28FIME%5BTIAB%5D%29+OR+%28TBC1+domain+family+member+24+isoform+1%5BTIAB%5D%29+OR+%28TBC1+domain+family+member+24+isoform+2%5BTIAB%5D%29+OR+%28skywalker+homolog%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- TBC1 DOMAIN FAMILY, MEMBER 24
<http://omim.org/entry/613577>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TBC1D24.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TBC1D24%5Bgene%5D>
- HGNC Gene Family: Deafness associated genes
<http://www.genenames.org/cgi-bin/genefamilies/set/1152>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=29203

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/57465>
- UniProt
<http://www.uniprot.org/uniprot/Q9ULP9>

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